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CASE REPORT

The role of ultrasound in the accurate diagnosis of a case of thanatophoric dysplasia

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Abstract Thanatophoric dysplasia (TD) has been reported as being one of the most common of the lethal skeletal dysplasias with its sonographic appearances being well-documented.

This report presents a case of TD, which was correctly diagnosed at a routine 20-week gestation anomaly ultrasound investigation. Full consent from the affected mother has been granted. Recent advances in ultrasound technologies have been shown to have had an impact on the detection of skeletal dysplasia. These advances, and the limitations of making an accurate diagnosis from ultrasound alone, are discussed within this report.

Introduction

Thanatophoric dysplasia (TD) is a lethal, rare osteochondrodysplasia[12] which was first described in 1967.[2] The term originates from the Greek for ''death bearing''. It is the most common of the lethal skeletal dysplasias with an incidence of 1 in 50,000 pregnancies within Europe.[3] TD results from mutations on the fibroblast growth factor receptor 3 (FGFR3) gene, on the short arm of chromosome 4.[4,12] Postpartum neonatal death occurs usually secondary to pulmonary hypoplasia caused by a small thoracic cavity.[1,3,5]

TD is characterised by extremely short ribs, a narrow thorax, markedly flattened vertebral bodies, short pelvis, platyspondyly (i.e. greatly reduced height of vertebral bodies and wide intervertebral spaces) prominent eyes, hypertelorism/euryopia (i.e. increased intra orbital distance) and depressed nasal bridge.[1–3,6] TD is subdivided into two types dependent on clinical features (Table 1); TD type I (TD1) is the most common accounting for 86% of cases.[6,7] These distinct features allow reliable radiologic and prenatal two-dimensional (2D) sonographic assessment.[7]

This case of thanatophoric dysplasia was diagnosed in the 20th week (20/40) of pregnancy by means of routine ultrasound examination.

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Table 1 The subdivision of TD on clinical characteristics

Characteristics	TD1	TD2
Long bones	Curved/bowed ''telephone receivers''	Straight
Skull	Mild lobulation of base of skull	Marked lobulation ''cloverleaf'' shape

Case report

A 35-year-old female (Gravida 3, Parity 1) attended for a routine 20/40 (2nd trimester) anomaly ultrasound scan. Measurements taken at a first trimester booking scan had agreed with menstrual dates and had found no obvious abnormalities. There was no known family history of any fetal anomalies.

The patient was examined using the GE Logic 9 imaging system with a multi-frequency (2.5—4 MHz) curvi-linear transducer (GE Medical Systems, Wisconsin, USA). A single fetus with a regular heartbeat was noted. All long bones in upper and lower extremities appeared bowed (Figs. 1 and 2) and markedly shortened for this gestational age[8,9] (Table 2). The thoracic circumference was markedly reduced compared to the abdominal circumference resulting in a ''champagne cork appearance'' (Fig. 3).[10] The fetal head was abnormally shaped with both a ''strawberry'' shape (Fig. 4) (in transverse section)[11] and frontal bossing (Fig. 3) (sagittal section) being evident.[6,7] A diagnosis of thanatophoric dysplasia was made.[6,7] Keeping to departmental protocols the lady was referred to the regional fetal medicine unit for further management. A tertiary level examination concurred with the earlier diagnosis of a lethal skeletal dysplasia, possibly thanatophoric.

Consent to a medical termination of pregnancy was obtained which was followed by a post-mortem [to include genetic assessment and bone biopsy].[7]

Pathologic findings included short symmetrical limbs with a bowed appearance, a narrow chest, frontal bossing and a depressed nasal bridge.[6,7] Histology of the bone showed severely disorganised cartilaginous tissue at the osteo-chondral growth zone.[4] Radiology examination reported rhizomelic deformity of the upper and lower limbs (i.e. the proximal long bones were shortened). Shortening of the ribs and prominent platyspondyly were also found[7] with the intervertebral disc space being about three times the height of the vertebral body (Fig. 5). Post-mortem findings confirmed the ultrasound diagnosis of thanatophoric dysplasia (TD1).

Discussion

TD is the most common form of skeletal dysplasia[1,3,5—7,10] and is due to autosomal dominant sporadic de novo mutations in FGFR3 gene mapped chromosome band 4p16.3.[4,10] The effects of this gene are expressed mainly in the skeletal and brain tissues of the developing fetus, where it is involved in growth regulation.[12,13] This leads to generalised defects and lack of endochondral ossification[4,7,11,13] both of which were evident in this case.

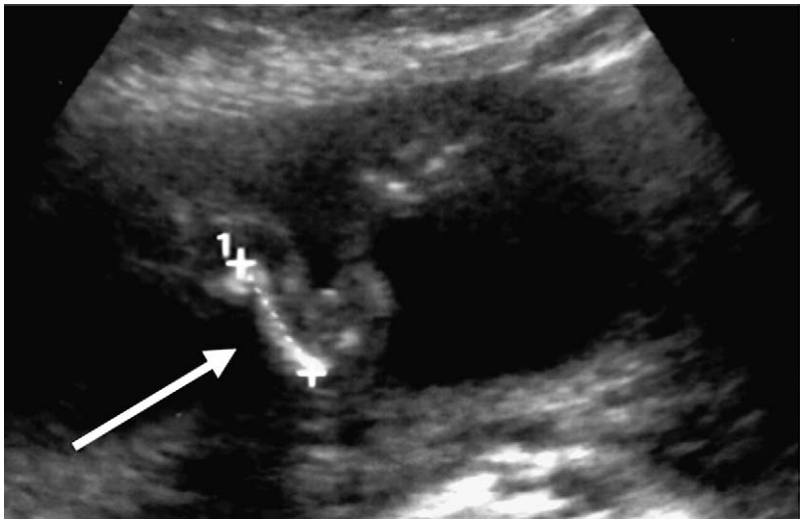

Figure 1 Long axis view of fetal arm, exhibiting extreme shortening of long bones. NB: humerus appears shortened and slightly bowed (arrow).